dietitian (RD) to help them learn how to manage their baby's diet.

Following the diet very strictly will give the baby with PKU the best chance to grow up healthy.

For more information, please contact:
South Carolina Department of Health and Environmental Control
Division of Women and Children's Services
Box 101106
Columbia, SC 29211
(803) 898-0767
or
your county health department

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Division of Women and Children's Services

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NEWBORN SCREENING 2

What You Should Know When a Second Test for PKU Is Needed

Newborn Screening

A small sample of your baby's blood was collected soon after birth and was sent to the DHEC laboratory for testing. This testing is called Newborn Screening. In SC, newborns are tested for several genetic and chemical disorders. These disorders include Phenylketonuria (called PKU), Congenital Hypothyroidism, Galactosemia, Congenital Adrenal Hyperplasia (called CAH), Medium Chain Acyl Co-A Dehydrogenase Deficiency (called MCADD), and Hemoglobinopathies. In some cases, a second test is needed to help your doctor decide if your baby has one of these disorders. In many cases, the second test will be normal. However, if your baby does have one of the newborn screening disorders, early treatment will give him or her the best chance to grow up healthy.

Phenylketonuria

Your baby's first test showed that he or she could possibly have Phenylketonuria. Here's a brief description of Phenylketonuria and how it is treated.

PKU is a genetic disorder that is found in around one of every 12,000 babies born each year. When a baby has PKU, he or she cannot break down phenylalanine, a part of the protein that is found in foods, including breast milk and infant formula.

If he or she is not treated, phenylalanine, or "phe," builds up in the baby's blood and damages the baby's brain causing mental retardation. It can also cause skin and nerve problems. Babies with untreated PKU will often have an unpleasant, musty odor.

Newborn screening allows the baby's doctor to tell if the baby probably has PKU before it causes irreversible damage to his or her brain. This lets the doctor give the parents special instructions on how to help the baby grow and develop normally.

Most babies drink either breast milk or infant formula as their main "food." Both breast milk and infant formula contain too much "phe" for a baby with PKU. A baby with PKU must have a special formula that limits the amount of "phe" that he or she eats each day.

As the baby begins to eat solid foods, the parents will have to be careful about which foods are given to the baby. The baby must not eat foods that contain a lot of "phe." As the baby grows up, he or she will need to drink a special formula and watch what is eaten very carefully throughout life.

The baby's doctor will usually refer the parents to a registered